

A B S T R A C T

Title: Methods

This invention relates to polymorphisms and novel sequence in the human pyruvate
5 dehydrogenase E1 α (PDH E1 α) gene. The invention also relates to methods and materials for
analysing allelic variation in the PDH E1 α gene, and to the use of PDH E1 α polymorphism in
the diagnosis and treatment of diseases in which modulation of pyruvate dehydrogenase
activity could be of therapeutic benefit, such as diabetes, asthma, obesity, sepsis and
peripheral vascular disease. In particular, the invention is based on the discovery of a
10 nucleotide polymorphism in the 3' untranslated region (3'UTR) of the human PDH E1 α gene.
In addition, we disclose the sequence of intron 7 of the human PDH E1 α gene and identify
two polymorphisms within intron 7.